

## Case Report

# “Leopard skin” sign: Magnetic resonance imaging appearance of Lhermitte-Duclos disease with histopathologic correlation

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## ABSTRACT

Dysplastic cerebellar gangliocytoma or Lhermitte-Duclos disease (LDD) is a rare hamartomatous malformation of cerebellum that can occur in isolation or as a component of Cowden syndrome. Although uncommon, the characteristic clinical symptoms and magnetic resonance imaging (MRI) features are highly suggestive of the condition and histopathological features confirm the diagnosis. We report a case of LDD, presenting with “Leopard skin” appearance of the lesion on MRI with a brief review of imaging, clinical, and histopathological features.

**Keywords:** Lhermitte-Duclos disease, Dysplastic cerebellar gangliocytoma, Magnetic resonance imaging, Leopard skin sign

## INTRODUCTION

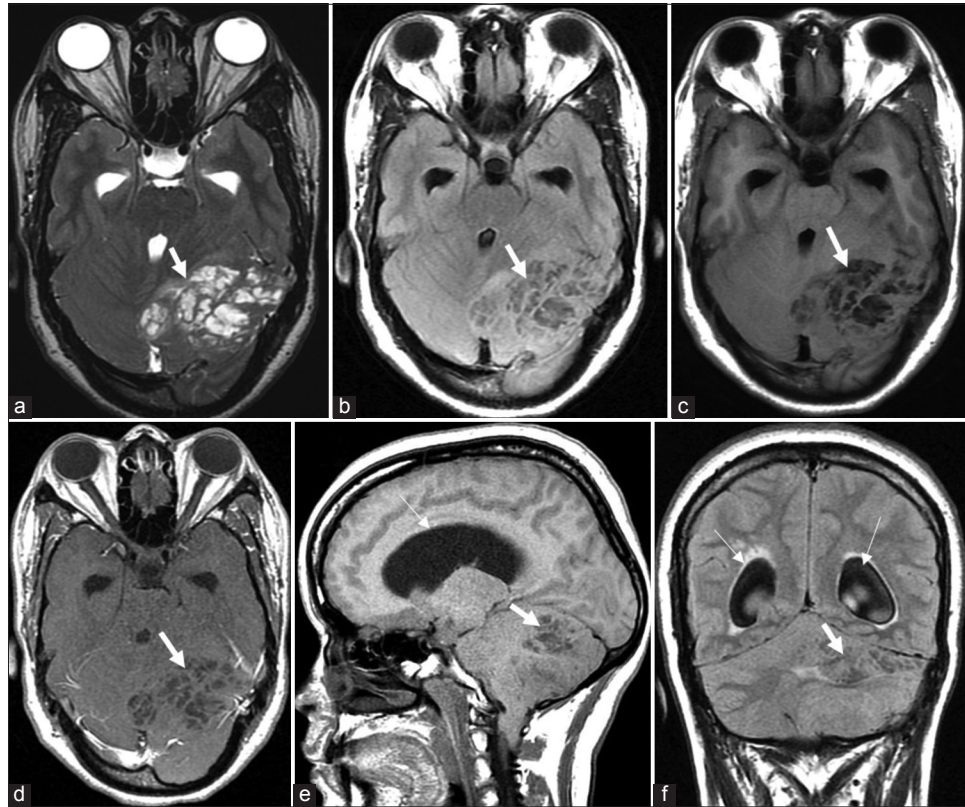
Dysplastic cerebellar gangliocytoma is a rare cerebellar hamartomatous lesion with characteristic imaging features. We report a case of LDD presenting with signs of raised intracranial tension.

## CASE REPORT

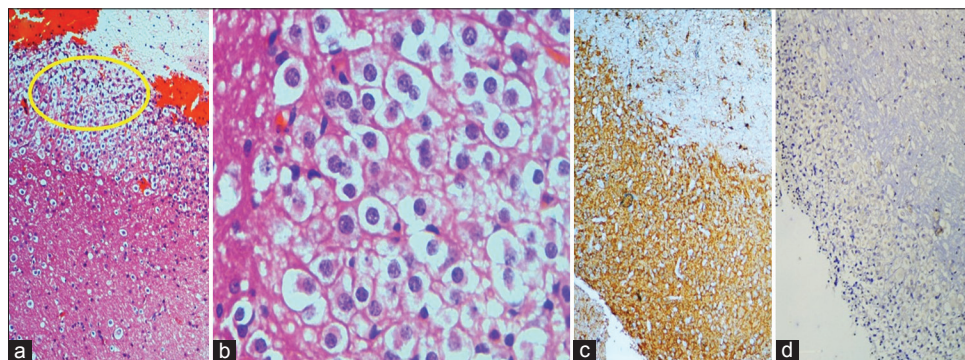
A 27-year-old female with a gradually progressive history of headache, giddiness, and vomiting of 2-month duration underwent evaluation with magnetic resonance imaging (MRI) at our institution. MRI of brain showed an ill-defined intra-axial lesion in the left cerebellar hemisphere with internal cystic foci and striations that are heterogeneously hypointense on T1 and hyperintense on T2-weighted images giving “leopard skin” appearance. Enhancement was not seen on contrast images and magnetic resonance spectroscopy was inconclusive. Mass effect with effacement of fourth ventricle and associated hydrocephalous was also noted [Figure 1]. Based on the location of lesion and the characteristic imaging findings, it was diagnosed as Lhermitte-Duclos disease (LDD). Surgery was performed with excision of lesion and histopathology confirmed the diagnosis [Figure 2]. Post-surgery, the symptoms have resolved and the patient is advised further follow-up.

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**Figure 1:** (a-c) Axial T2-weighted image, T2-weighted-fluid-attenuated inversion recovery (T2 FLAIR), and T1-weighted image (T1W) magnetic resonance imaging images show unilateral cerebellar hypertrophy exhibiting the characteristic “leopard skin” appearance with alternating hypointense and hyperintense cystic areas and striations (thick arrows). (d) T1W contrast image shows non-enhancement of the lesion. (e and f) Sagittal T1W and coronal T2 FLAIR images show the dilated lateral ventricles (thin arrows) secondary to the lesion (thick arrows) causing obstructive hydrocephalus.



**Figure 2:** H&E staining of tumor tissue (a). Shows diffuse enlargement of the molecular and internal granular layers with abnormally myelinated axon bundles in outer molecular layer (magnification  $\times 10$ ). (b) Dysplastic granular neurons shown in the inset of (a) (magnification  $\times 40$ ). (c and d) Immunohistochemistry positive for Synaptophysin and Ki67.

## DISCUSSION

Dysplastic cerebellar gangliocytoma or LDD, named after French physicians “Jacques Jean Lhermitte” and “P Duclos,” is

an uncommon hamartomatous malformation of cerebellum classified under central nervous system World Health Organization Grade 1 glioneuronal and neuronal tumors.<sup>[1]</sup>

**Table 1:** Differential diagnosis of unilateral cerebellar lesions.

Cerebellar lesion	Clinical features	MRI features
Lhermitte-Duclos-Disease	Dysplastic cerebellar gangliocytoma, usually seen in adults, association with Cowden's syndrome	Alternating hypointense and hyperintense linear and rounded areas with typical "tigroid" or "Leopard skin" appearance
Cerebellar cortical dysplasia/heterotopias	Focal area of disorganized cerebellar architecture, association with chromosomal abnormalities, intrauterine infections, and congenital muscular dystrophies	Folial thickening, irregular grey-white matter interface, abnormal fissures, and cortical cystic lesions
Acute cerebellitis	Secondary to acute infection or inflammation, most commonly of viral etiology	Rarely unilateral. T2 hyperintense lesion with perilesional edema, mild diffusion restriction, and contrast-enhancement with or without adjacent leptomeningeal involvement
Sonic hedgehog activated medulloblastoma	Originate from the cerebellar granule neuron progenitor lineage and usually occur in the lateral parts of the cerebellar hemispheres	Isointense on T1 and T2 W sequences with cystic areas, perilesional edema, diffusion restriction, and contrast-enhancement
High grade neuroectodermal tumor	Associated with <i>BCOR</i> gene exon 15 internal tandem duplication	Large, well-circumscribed, heterogeneous masses with variable enhancement and reduced diffusion

MRI: Magnetic resonance imaging

The incidence of this condition is rare with around 230 cases reported in the literature. LDD can occur either in isolation or as a component of Cowden syndrome associated with *PTEN* mutations. It is usually seen in adults without any race or gender predilection. Clinical symptoms depend on the size of the lesion and patients with small lesions can be asymptomatic. Patients with large lesions present with signs and symptoms of cerebellar dysfunction (vertigo and ataxia) and raised intracranial pressure due to mass effect (headache, vomiting, cranial nerve palsies, and hydrocephalus).<sup>[2]</sup> An extremely rare case of LDD presenting with psychiatric disturbances was also reported.<sup>[2]</sup>

On MRI, LDD is typically seen as an expansile unilateral cerebellar lesion with alternating hypointense and hyperintense linear and rounded areas giving rise to the typical "tigroid" pattern of appearance or the less common "Leopard skin" appearance. The lesion does not show any significant contrast enhancement or diffusion restriction.

On histopathology, macroscopically, the lesion is characterized by cerebellar folial hypertrophy and thinning of medullary white matter. On microscopic examination, the lesion characteristically shows loss of granular cells and purkinje cells with an overgrowth of dysplastic ganglionic cells.

The differential diagnosis of LDD with clinical and imaging features is given in Table 1.<sup>[3]</sup>

Asymptomatic patients can be left alone, whereas, symptomatic larger lesions causing mass effect need surgical resection.<sup>[4]</sup> Although complete surgical resection is not possible in all the cases and recurrence can occur after surgery, prognosis is favorable due to the slow growth and progression of the lesion.<sup>[4,5]</sup>

## CONCLUSION

LDD should be considered as a differential diagnosis in young adults presenting with signs and symptoms of unilateral cerebellar mass and raised intracranial hypertension.

## LEARNING POINTS

- Although the incidence of LDD is extremely rare, clinical symptoms related to the location of the lesion and the characteristic "tigroid" or "leopard skin" MR imaging appearance are diagnostic of this condition, and the typical histopathologic features confirm the diagnosis.
- LDD occurring as a part of Cowden syndrome (multiple hamartoma syndrome) is associated with increased risk of melanomas, breast, endometrium, thyroid, and colorectal malignancies; hence, further clinical assessment and radiological screening are recommended in these patients.

## MCQs

- The most common malignancy associated with Cowden's syndrome is
  - Ductal breast cancer
  - Dysplastic gangliocytoma of the cerebellum
  - Colorectal cancer
  - Melanoma

Answer Key: a

- <sup>18</sup>F-FDG PET/CT scan in LDD shows
  - Increased uptake in all lesions
  - Increased uptake in the vicinity of the lesions
  - Increased uptake in and around the lesions

- d. Variable uptake in and around the lesion, not specific for the diagnosis

Answer Key: d

3. In regard to calcifications in the lesions of LDD, the following statement is correct
- Calcifications are seen in all the lesions
  - Calcifications are seen in few cases
  - Lesions never exhibit calcifications
  - None of the above

Answer Key: b

#### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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#### Conflicts of interest

There are no conflicts of interest.

#### Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the

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